

Variant: *NM_001110792.2(MECP2):c.800G>C (p.Arg267Pro)*

Version: 1.0

CA415171753 [↗](#)

2229884 (ClinVar) [↗](#)

Gene: MECP2 ([HGNC:4204](#))

Condition: Rett syndrome ([MONDO:0010726](#))

Inheritance Mode: X-linked inheritance

UUID: 6708388a-56c1-4c91-8b7f-c85de294d2ad

Approved on: 2026/1/28

Published on: 2026/3/31

HGVS expressions

NM_001110792.2(MECP2):c.800G>C (p.Arg267Pro)

NM_001110792.2:c.800G>C

NC_000023.11:g.154031064C>G

CM000685.2:g.154031064C>G

NC_000023.10:g.153296515C>G

CM000685.1:g.153296515C>G

NC_000023.9:g.152949709C>G

NG_007107.2:g.111064G>C

NG_007107.3:g.111040G>C

ENST00000303391.11:c.764G>C

ENST00000453960.7:c.800G>C

ENST00000637917.1:c.66-128G>C

ENST00000303391.10:c.764G>C

ENST00000407218.5:c.*136G>C

ENST00000453960.6:c.800G>C

ENST00000619732.4:c.764G>C

ENST00000622433.4:c.752G>C

ENST00000628176.2:c.*136G>C

NM_001110792.1:c.800G>C

NM_001316337.1:c.485G>C

NM_004992.3:c.764G>C

NM_001316337.2:c.485G>C

NM_001369391.2:c.485G>C

NM_001369392.2:c.485G>C

NM_001369393.2:c.485G>C

NM_001369394.1:c.485G>C

NM_001369394.2:c.485G>C

NM_001386137.1:c.95G>C

NM_001386138.1:c.95G>C

NM_001386139.1:c.95G>C

NM_004992.4:c.764G>C

Pathogenic

Met criteria codes **4**

PM2_Supporting

PS4_Moderate

PS2_Very Strong

PM6

Expert Panel

Rett and Angelman-like Disorders VCEP [↗](#)

Not Met criteria codes 1

PP3

Evidence Links 0

Criteria Specification Information

[Criteria Specification:](#) *ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MECP2 Version 5.0.0*

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Arg255Pro variant in MECP2 (NM_004992.3) has been reported as a de novo occurrence (biological parentage both confirmed and unconfirmed) in at least 3 individuals with features of Rett syndrome (Ambry Genetics- internal database, GeneDx- internal database) (PS2_Very strong). The p.Arg255Pro variant in MECP2 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with features of Rett syndrome (GeneDx- internal database) (PM6). The p.Arg255Pro variant has been observed in 3 individuals with affected with a neurodevelopmental phenotype consistent with MECP2-related conditions (Labcorp Genetics (formerly Invitae)- internal database; Ambry Genetics- internal database) (PS4_moderate). The p.Arg255Pro variant in MECP2 is absent from gnomAD v4.1 (PM2_Supporting). Computational prediction analysis tools are inconclusive for this variant. In summary, the p.Arg255Pro variant in MECP2 is classified as Pathogenic based on the ACMG/AMP criteria (PS2_very-strong, PM6, PS4_moderate, PM2_Supporting). (MECP2 Specifications v.5.0; curation approved on 01/28/2026)

Met criteria codes

PM2_Supporting

i

✓

The p.Arg255Pro variant in MECP2 is absent from gnomAD v4.1 (PM2_Supporting).

PS4_Moderate

i

✓

The p.Arg255Pro variant has been observed in 3 individuals with affected with a neurodevelopmental phenotype consistent with MECP2-related conditions (Labcorp Genetics (formerly Invitae)- internal database; Ambry Genetics- internal database) (PS4_moderate).

PS2_Very Strong

i

✓

The p.Arg255Pro variant in MECP2 has been reported as a de novo occurrence (biological parentage confirmed) in at least 2 individuals with features of Rett syndrome (Ambry Genetics- internal database) (PS2_Very strong).

PM6

i

✓

The p.Arg255Pro variant in MECP2 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with features of Rett syndrome (GeneDx- internal database) (PM6).

Not Met criteria codes

PP3

i

✗

REVEL = 0.617. Computational prediction analysis tools are inconclusive for this variant.

Curation History [↗](#)

▾

▾

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.